Summary of Core Recommendations: Myotonic Dystrophy CDEs

Start-up Resource – NINDS Myotonic Dystrophy Disease CDE Recommendations

The National Institute of Neurological Disorders and Stroke (NINDS) and other Federal agencies and international organizations have the common mission of developing data standards for clinical research. Through the efforts of subject-specific working groups, topic-driven data elements have been created. The first set of Common Data Elements (CDEs) for Myotonic Dystrophy was developed in 2014. The Core data elements to be used by an investigator when beginning a research study in this disease/disorder are listed in this resource document. All other recommendations are listed on the website and should be considered based on study type.

Each CDE or instrument could be classified according to the definitions below:

General Core: A data element that is required for all NINDS funded studies.

Disease Core: A data element that collects essential information applicable to any disease-specific study, including all therapeutic areas. The NINDS and its appointed working groups assign the disease "Core" classification based on the current clinical research best practices. In each case, the disease Core CDEs are a small subset of the available CDEs, where it is anticipated that investigators will need to collect the disease Core CDEs on any type of study. These are required for all disease-specific studies.

Disease Supplemental - Highly Recommended: A data element which is essential based on certain conditions or study types in clinical research studies. In most cases, these have been used and validated in the disease area. These data elements are strongly recommended for the specified disease condition, study type or design.

Disease Supplemental: A data element which is commonly collected in clinical research studies. Use depends upon the study design, protocol or type of research involved. These are recommended, but not required, for studies.

Disease Exploratory: A data element that requires further validation but may fill current gaps in the CDEs and/or substitute for an existing CDE once validation is complete. Such data elements show great promise but require further validation before they are ready for prime-time use in clinical research studies. They are reasonable to use with the understanding that it has limited validation in the target group.

National Institute of Health (NIH) Resources:	
The NINDS also strongly encourages researchers to use these NIH developed materials for NINDS-sponsored research, when appropriate. Utilization of these resources will enable greater consistency for NINDS-sponsored research studies. These tools are free of charge.	• **NIH Toolbox** • **Quality of Life in Neurological Disorders (Neuro-QOL)** • **Patient-Reported Outcomes Measurement Information System (PROMIS)**

Core CDEs for all NINDS Studies[1]:

Domain/Sub-Domain	CDE Name	CDE ID	Study Type
Participant Characteristics; Demographics	Birth date	C00007	All studies
Participant Characteristics; Demographics	Ethnicity USA category	C00020	All studies
Participant Characteristics; Demographics	Race USA category	C00030	All studies
Participant Characteristics; Demographics	Birth sex assigned type	C58676	All studies
Participant Characteristics; Demographics	Gender identity type	C58677	All studies
Participant History and Family History; General Health History	Medical history condition text	C00322	All studies
Participant History and Family History; General Health History	Medical history condition SNOMED CT code	C00313	All studies

General Core for all Studies:

Investigators should review the FDA's "Guidance for Industry: Suicidal Ideation and Behavior: Prospective Assessment of Occurrence in Clinical Trials" for the most up-to-date information about suicidal ideation and behavior. One scale that FDA suggests is the Columbia Suicide Severity Rating Scale (C-SSRS) (available at Columbia Suicide Severity Rating Scale).

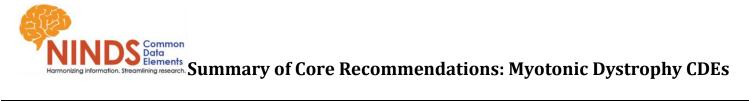[1] Note: Education year count C00015 is no longer a general Core CDE

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Core CDEs for Myotonic Dystrophy:

Domain; Sub-Domain	Data Element	CDE ID
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Specimen source type	C12226
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Tissue specimen size measurement	C12229
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Tissue specimen collection date and time	C12230
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Histochemical stains used other text	C18828
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Biopsy and autopsy fiber abnormality type	C12525

Supplemental-Highly Recommended CDEs for Myotonic Dystrophy:

Domain; Sub-Domain	Data Element	CDE ID
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Preservation technique used type	C12233
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Histochemical stains used type	C12235

Domain; Sub-Domain	Data Element	CDE ID
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Biopsy and autopsy histochemical stains diagnostic abnormalities present type	C12249
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Biopsy and autopsy type 1 predominance fiber percentage value	C12526
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Biopsy and autopsy type 2 predominance fiber percentage value	C12527
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Lab test other text	C18731
Assessments and Examinations; Non-Imaging Diagnostics	ECG administer date time clock type	C17967
Assessments and Examinations; Non-Imaging Diagnostics	ECG atrial rate	C12823
Assessments and Examinations; Non-Imaging Diagnostics	ECG PR interval	C04509
Assessments and Examinations; Non-Imaging Diagnostics	ECG QRS duration	C04510
Assessments and Examinations; Non-Imaging Diagnostics	ECG result text	C17966
Assessments and Examinations; Non-Imaging Diagnostics	ECG arrhythmia ventricular morphology type	C12826
Assessments and Examinations; Non-Imaging Diagnostics	ECG pace rhythm type	C12825
Assessments and Examinations; Non-Imaging Diagnostics	ECG axis direction indicator	C12827

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Domain; Sub-Domain	Data Element	CDE ID
Assessments and Examinations; Non-Imaging Diagnostics	ECG atrial enlargement right indicator	C12828
Assessments and Examinations; Non-Imaging Diagnostics	ECG atrial enlargement left indicator	C12829
Assessments and Examinations; Non-Imaging Diagnostics	ECG WPW syndrome indicator	C10758
Assessments and Examinations; Non-Imaging Diagnostics	ECG conduction block indicator	C12830
Assessments and Examinations; Non-Imaging Diagnostics	ECG atrioventricular block first degree indicator	C12831
Assessments and Examinations; Non-Imaging Diagnostics	ECG atrioventricular block second degree indicator	C12832
Assessments and Examinations; Non-Imaging Diagnostics	ECG atrioventricular block third degree complete indicator	C12833
Assessments and Examinations; Non-Imaging Diagnostics	ECG complete bundle branch block indicator	C04507
Assessments and Examinations; Non-Imaging Diagnostics	ECG bundle branch block incomplete indicator	C12834
Assessments and Examinations; Non-Imaging Diagnostics	ECG bundle branch block type	C12835
Assessments and Examinations; Non-Imaging Diagnostics	ECG left anterior hemiblock indicator	C12836
Outcomes and Endpoints; Pulmonary Function Testing/Respiratory Status	Pulmonary function test date and time	C11098

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Domain; Sub-Domain	Data Element	CDE ID
Outcomes and Endpoints; Pulmonary Function Testing/Respiratory Status	Pulmonary function test not done indicator	C17874
Outcomes and Endpoints; Pulmonary Function Testing/Respiratory Status	Pulmonary function test not done reason	C12298
Outcomes and Endpoints; Pulmonary Function Testing/Respiratory Status	Pulmonary function test type	C11099
Outcomes and Endpoints; Pulmonary Function Testing/Respiratory Status	Pulmonary function test not done text	C17876
Outcomes and Endpoints; Pulmonary Function Testing/Respiratory Status	Pulmonary function test position type	C11100
Outcomes and Endpoints; Pulmonary Function Testing/Respiratory Status	Pulmonary Function Test seat position type	C12301
Outcomes and Endpoints; Pulmonary Function Testing/Respiratory Status	Pulmonary function test mouth apparatus type	C11101
Outcomes and Endpoints; Pulmonary Function Testing/Respiratory Status	Pulmonary function test equipment manufacturer name	C12303
Outcomes and Endpoints; Pulmonary Function Testing/Respiratory Status	Pulmonary function test equipment model name	C12304

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Domain; Sub-Domain	Data Element	CDE ID
Outcomes and Endpoints; Pulmonary Function Testing/Respiratory Status	Pulmonary function test equipment software program name	C12305
Outcomes and Endpoints; Pulmonary Function Testing/Respiratory Status	Ulna length measurement	C12306
Outcomes and Endpoints; Pulmonary Function Testing/Respiratory Status	Ulna length measure tool name	C12307
Outcomes and Endpoints; Pulmonary Function Testing/Respiratory Status	Pulmonary function test bronchodilator indicator	C12308
Outcomes and Endpoints; Pulmonary Function Testing/Respiratory Status	Trial number	C10171
Outcomes and Endpoints; Pulmonary Function Testing/Respiratory Status	Pulmonary function complete exhalation indicator	C12310
Outcomes and Endpoints; Pulmonary Function Testing/Respiratory Status	Pulmonary function index of lung function type	C12311
Outcomes and Endpoints; Pulmonary Function Testing/Respiratory Status	Pulmonary function lung function measurement	C12516
Outcomes and Endpoints; Pulmonary Function Testing/Respiratory Status	Pulmonary function index of lung function best trial measurement	C12312

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Domain; Sub-Domain	Data Element	CDE ID
Outcomes and Endpoints; Pulmonary Function Testing/Respiratory Status	Pulmonary function maximal pressure trial difference indicator	C12313
Outcomes and Endpoints; Pulmonary Function Testing/Respiratory Status	Pulmonary function forced vital capacity cough exhalation indicator	C12314
Outcomes and Endpoints; Pulmonary Function Testing/Respiratory Status	Pulmonary function forced vital capacity leak indicator	C12315
Outcomes and Endpoints; Pulmonary Function Testing/Respiratory Status	Pulmonary function forced vital capacity large value range indicator	C12316
Outcomes and Endpoints; Pulmonary Function Testing/Respiratory Status	Pulmonary function forced vital capacity early termination indicator	C12317
Outcomes and Endpoints; Pulmonary Function Testing/Respiratory Status	Pulmonary function forced vital capacity peak flow indicator	C12318

Supplemental Highly Recommended Instruments for Myotonic Dystrophy:

1. Manual Muscle Testing-Using the Medical Research Council Muscle Grading Scale

2. Bedside Myotonia

3. Epworth Sleepiness Scale

4. Myotonic Dystrophy Health Index (MDHI)

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For the complete list of NINDS CDE recommendations for DM, please see the [NINDS CDE website](#).